

Hybrid event



11th International Conference on Rare and Undiagnosed Diseases

7th – 8th November 2022
Van Swieten Saal, Medical University of Vienna
Van-Swieten-Gasse 1a, 1090 Vienna

www.meduniwien.ac.at/udni

11th International Conference on Rare and Undiagnosed Diseases

Rare diseases continue to pose enormous challenges for clinicians, healthcare institutions, society, and the daily life of affected individuals. Sometimes patients are even left without a defined diagnosis (undiagnosed diseases). While impressive achievements have elucidated the underlying pathomechanisms of rare diseases, several barriers need to be overcome in order for patients to benefit from recent research advances.

Vienna has become a hotspot for rare and undiagnosed disease research in Europe, including activities of the Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD) and its prestigious partner institutions, the Medical University of Vienna, St. Anna Children's Cancer Research Institute and the CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences. Beyond institutional, local and national boundaries, important alliances have been formed, including a prioritized partnership with the NIH Undiagnosed Diseases Program and the Undiagnosed Diseases Network International (UDNI).

We are honored to host this 11th Conference of the Undiagnosed Diseases Network International (UDNI) jointly with the NIH and the Medical University of Vienna. We welcome congress participants who will jointly shape the future of rare disease research and clinical translation and also critically review and overcome the current societal challenges of rare diseases.

We are looking forward to seeing you at the meeting and to engage in many active and fruitful discussions.

Kaan Boztug, Georg Stary, William A. Gahl

Conference Chairs

Kaan Boztug, Medical University of Vienna, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, St. Anna Children's Cancer Research Institute (CCRI), St. Anna Children's Hospital, Austria

Georg Stary, Medical University of Vienna, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, Austria

William A. Gahl, National Institutes of Health (NIH), USA

Invited Speakers Day 1

(listed in chronological order)

- Alan Warren**, University of Cambridge, UK
- Anne Puel**, IMAGE Institute, France
- Veronica Kinsler**, University College London GOS Institute of Child Health, UK
- Aleixo Muise**, SickKids-Hospital for Sick Children, Canada
- Gaia Novarino**, Institute of Science and Technology (IST), Austria
- Han Brunner**, Radboud University Medical Center, Maastricht University Medical Center, Netherlands
- Camilo Toro**, National Institutes of Health (NIH), USA
- Jörg Menche**, University of Vienna and Max Perutz Labs, Austria
- Lukas Huber**, Medical University of Innsbruck, Austria
- Alessandra Biffi**, University of Padua, Italy

Organizing Committee

Christoph Bock, CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, Medical University of Vienna, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), Austria

Christiane Druml, Medical University of Vienna, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), Austria

Loïc Dupré, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), Austria

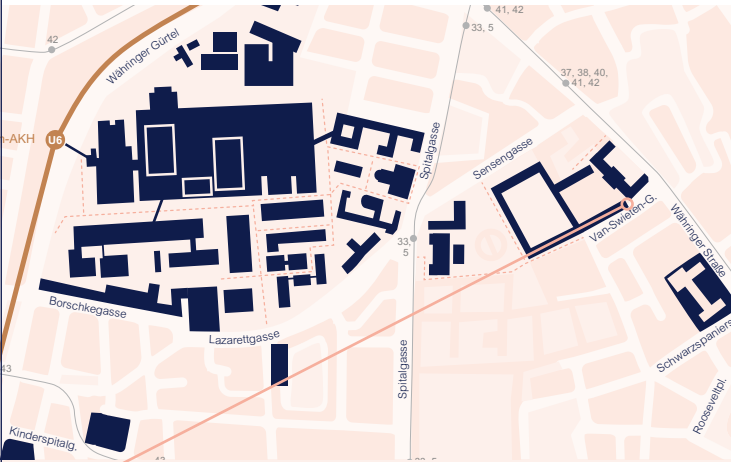
Vanja Nagy, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), Austria

Barbara Prainsack, University of Vienna, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), Austria

Thomas Reiberger, Medical University of Vienna, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, Austria

Andreas Villunger, Medical University of Innsbruck, Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD), CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, Austria

Registration under: www.udninternational.org



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1090 Vienna

In cooperation with



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Programme, 7th November

12.00 – 12.20 pm
Welcome and Opening
Markus Müller, Rector, MedUni Vienna, Austria
Giulio Superti-Furga, Scientific Director, CeMM, Austria
Kaan Boztug, MedUni Vienna/LBI-RUD/CeMM/St. Anna CCRI and St. Anna Children’s Hospital, Austria
Georg Stary, MedUni Vienna/LBI-RUD/CeMM, Austria
William A. Gahl, National Institutes of Health (NIH), USA
Helene Cederroth, Wilhelm Foundation, Sweden

**Session 1:
Immunodeficiencies and hematological diseases**

Chair: Marta Rizzi, MedUni Vienna, Austria and University of Freiburg, Germany
Leo Kager, St. Anna Children’s Hospital, Austria

12.20 – 12.50 pm
New rare disease entities at the interface of immunodeficiency, immune dysregulation and cancer predisposition
Kaan Boztug, MedUni Vienna/LBI-RUD/CeMM/St. Anna CCRI and St. Anna Children’s Hospital, Austria

12.50 am – 1.20 pm
Convergent somatic evolution commences in utero in a germline ribosomopathy
Alan Warren, University of Cambridge, UK

1.20 – 1.50 pm
Rare diseases shed light in the middle of a global health crisis: Human host factors in critical COVID-19 pneumonia
Anne Puel, IMAGE Institute, France

1.50 – 2.00 pm
Selected oral abstract 1

2.00 – 2.10 pm
Selected oral abstract 2

**Session 2:
Dermatology plus Gastroenterology**

Chair: Wolfgang Weninger, MedUni Vienna, Austria
Loïc Dupré, LBI-RUD, Austria

2.10 – 2.40 pm
Birthmarks – unravelling the causes, understanding the implications
Veronica Kinsler, University College London GOS Institute of Child Health, UK

2.40 – 3.10 pm
The skin immune system in rare diseases
Georg Stary, MedUni Vienna/LBI-RUD/CeMM, Austria

3.10 – 3.40 pm
Novel monogenic intestinal disease: New genes leading to treatments
Aleixo Muise, SickKids–Hospital for Sick Children, Canada

3.40 – 3.50 pm
Selected oral abstract 1

3.50 – 4.00 pm
Selected oral abstract 2

4.00 – 4.30 pm
Coffee Break

**Session 3:
Neurological Diseases**
Chair: Vanja Nagy, LBI-RUD, Austria
Sofia Grade, IMBA, Austria

4.30 – 5.00 pm
Studying brain development through the lens of human rare diseases
Gaia Novarino, IST Austria

5.00 – 5.30 pm
A new look at recessive: Why do we have the diseases we have?
Han Brunner, Radboud UMC, Maastricht UMC+, Netherlands

5.30 – 6.00 pm
New and rare movement disorders in the NIH Undiagnosed Diseases Program
Camilo Toro, NIH, USA

6.00 – 6.10 pm
Selected oral abstract 1

6.10 – 6.20 pm
Selected oral abstract 2

**Session 4:
Model systems and Therapeutics**
Chair: Christoph Bock, CeMM/MedUni Vienna/LBI-RUD, Austria
Susanne Greber-Platzer, MedUni Vienna, Austria

6.20 – 6.50 pm
Network approaches for rare diseases
Jörg Menche, University of Vienna and Max Perutz Labs, Austria

6.50 – 7.20 pm
Congenital enteropathies: many mutations, different clinical presentation, but all always related to intracellular protein transport and sorting
Lukas Huber, MedUni Innsbruck, Austria

7.20 – 7.50 pm
HSC gene therapy for neurometabolic and neuro-degenerative diseases: current and new indications
Alessandra Biffi, University of Padua, Italy

7.50 – 8.00 pm
Selected oral abstract 1

8.00 – 8.10 pm
Selected oral abstract 2

8.10 – 8.25 pm
Break

8.25 – 8.55 pm
**Podium Discussion
The future of rare diseases – challenges and possible solutions**
William A. Gahl, National Institutes of Health (NIH), USA
Christiane Druml, MedUni Vienna/LBI-RUD, Austria
Michaela Fritz, Vice Rector, MedUni Vienna, Austria
Cornelia Dechant, Kardiologie Wieden, Austria
Helene Cederroth, Wilhem Foundation, Sweden
Moderation: Marlene Nowotny, ORF

8.55 – 9.05 pm
Final Remarks
Kaan Boztug, MedUni Vienna/LBI-RUD/CeMM/St. Anna CCRI and St. Anna Children’s Hospital, Austria
Georg Stary, MedUni Vienna/LBI-RUD/CeMM, Austria
William A. Gahl, National Institutes of Health (NIH), USA

9.05 – 10.30 pm
Poster Walk and Get together

8th November

12.00 – 7.30 pm
UDNI Committee and Working Group Reports
Chair: William A. Gahl, National Institutes of Health (NIH), USA
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